

RESEARCH SUMMARY

HEALTHCARE EXPERIENCES OF AFRICAN AMERICAN WOMEN WITH THE FRAGILE X PREMUTATION

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PURPOSE

The purpose of this study is to understand the healthcare experiences, family dynamics, and mental health of African American women with a fragile X premutation (PM).

BACKGROUND

Fragile X syndrome (FXS) is a genetic disorder caused by a mutation on the X-chromosome. Like other genetic disorders, people can be carriers of FXS, described as carrying a premutation (PM). But unlike most disorders where the carrier isn't affected, people with a fragile X PM are at risk to experience conditions associated with FXS such as fragile X-associated primary ovarian insufficiency (FXPOI), fragile X-associated tremor ataxia syndrome (FXTAS), and a group of mental health conditions called fragile X-associated neuropsychiatric disorders (FXAND).

Though a PM hasn't been indicated to affect women based on race/ethnicity, women of color have historically been underrepresented in research around FXS-associated conditions. Research has shown healthcare access and quality in America is influenced by race, with psychological, social, cultural, and structural factors that act as barriers for African American women. This study aims to provide insight on the healthcare experiences of African American women with a PM.

STUDY PARTICIPANTS

African American women who carry a PM were identified through the Fragile X Research Participant Registry of the Carolina Institute for Developmental Disabilities (CIDD) as well as the Emory Fragile X Center Registry.

METHOD

- An interview guide was developed
- In-depth phone interviews were conducted to explore healthcare experiences, the journey to genetic testing, the disclosure of participant's PM results, sources of support and follow-up care, changes of emotional health, and advice from participants.

THEMES FROM INTERVIEWS

Healthcare Experiences

Healthcare providers (HCPs) were perceived as supportive/informed when they were willing to acknowledge limited knowledge on fragile X PMs and make appropriate referrals. They were also seen as supportive if they educated themselves on fragile X PMs and worked with participants to make decisions for their care. Though knowledge and empathy aren't always related, one participant found support through the knowledge of a well-informed provider as well as an empathetic response from another provider.

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THEMES FROM INTERVIEWS (CONT.)

Healthcare Experiences (Cont.)

HCPs were perceived as unsupportive/uninformed when participants had to inform their provider on the fragile X PM and the provider was not willing to learn about fragile X and its associated conditions. Moderately informed doctors were also seen as less supportive as they could be misguiding. Though well-informed providers can be seen positively, a participant negatively perceived a response she found to be “cold” and “very technical” from a well-informed provider. Some participants also sought out female HCPs to receive a quality of care they did not find with their male providers.

Participants also faced external barriers to care such as issues with insurance and healthcare practices. For example, when a provider would leave a practice, some struggled to find continued care. Others found it hard to find informed HCPs when insurance changes took them to different practices. Participants also reported internal barriers such as worry that they weren't being taken seriously and mistrust of the medical institution.

Family Dynamics

Many participants had to inform family members of a child's FXS diagnosis in addition to their PM diagnosis. Though one participant depicted a supportive experience, all other participants described reactions that included denial, blaming others, avoiding blame, misattributing symptoms, and a lack of support. While some participants experienced closeness and support from their families, others felt that their diagnosis caused strain in their relationships. As many participants also had an affected child, they found themselves being advocates and protectors for their children in addition to the traditional roles of childcare.

Emotional & Mental Health

Some emotional states participants experienced related to diagnosis, navigating healthcare, and sharing information with family are trauma, validation, frustration, grief, anger, relief, fear, guilt, and isolation. Participants found support through connections such as partners, friends, educators, therapists, and employers. They also found support from in-person or online support groups and religious communities.

Participants were asked about mental health due to the potential for FXAND. Six participants had a diagnosis of depression or anxiety, and half the patients felt their symptoms were well-managed with medication.

Advice

Some advice participants had for HCPs is to be more empathetic and informed about the fragile X PM. They discussed the need for referrals, especially for parents when they are diagnosed with a PM through a FX diagnosis of a child. They suggested that intake material should include genetic conditions to minimize traumatic questions in healthcare experiences.

Advice given to national patient and research groups is to be more inclusive and supportive of single parents raising kids with FXS. One participant mentioned that she'd like to see more research on the transition into adulthood for those with FXS.

The advice the participants had for other African American women with a PM is to become self-advocates and to seek out educational materials. Half the participants suggested for women to get involved with research.

CONCLUSION

The healthcare experiences of African American women with a PM are impacted by the support and knowledge of their healthcare providers, external barriers like insurance, and internal barriers like distrust in the American medical system. More resources need to be dedicated towards understanding and addressing the needs of African American families in the fragile X community to improve care of African American women impacted by fragile X.